

Title (en)
PROTEOMIC SCREENING FOR DISEASES

Title (de)
PROTEOMISCHES SCREENING FÜR KRANKHEITEN

Title (fr)
DÉPISTAGE PROTÉOMIQUE POUR MALADIES

Publication
EP 4114851 A1 20230111 (EN)

Application
EP 21764238 A 20210303

Priority
• US 202062984744 P 20200303
• US 2021020679 W 20210303

Abstract (en)
[origin: WO2021178545A1] Clinical diagnosis and newborn screening for primary immunodeficiencies (PIDDs) is described. The PIDDs include X-linked chronic granulomatous disease (X-CGD), X-linked lymphoproliferative syndrome (XLP1; SH2D1A deficiency), familial hemophagocytic lymphohistiocytosis 2 (FHL2), ataxia telangiectasia (AT), common variable immunodeficiency (CVID; B-cell dysfunctions), severe combined immunodeficiency (SCID), adenosine deaminase (ADA) deficiency, and dedicator of cytokinesis 8 (DOCK8) deficiency. Additionally, cell specific markers for platelets (CD42), natural killer (NK) cells (CD56), and T cells (CD3epsilon and CD3delta) can be used as secondary markers to provide support for diagnosis of disease.

IPC 8 full level
C07K 14/705 (2006.01); **C07K 14/725** (2006.01); **C07K 16/28** (2006.01); **G01N 33/577** (2006.01)

CPC (source: EP US)
C07K 16/18 (2013.01 - EP); **C07K 16/2803** (2013.01 - EP); **C07K 16/2896** (2013.01 - EP); **C07K 16/40** (2013.01 - EP); **C12N 9/0036** (2013.01 - EP); **C12Y 106/03001** (2013.01 - EP); **G01N 33/6848** (2013.01 - EP); **G01N 33/6893** (2013.01 - EP US); **C07K 2317/34** (2013.01 - EP); **G01N 2333/70503** (2013.01 - EP); **G01N 2333/70596** (2013.01 - EP); **G01N 2570/00** (2013.01 - EP US); **G01N 2800/24** (2013.01 - EP US); **G01N 2800/52** (2013.01 - EP)

Designated contracting state (EPC)
AL AT BE BG CH CY CZ DE DK EE ES FI FR GB GR HR HU IE IS IT LI LT LU LV MC MK MT NL NO PL PT RO RS SE SI SK SM TR

Designated extension state (EPC)
BA ME

Designated validation state (EPC)
KH MA MD TN

DOCDB simple family (publication)
WO 2021178545 A1 20210910; CN 115279783 A 20221101; EP 4114851 A1 20230111; EP 4114851 A4 20240717; JP 2023516733 A 20230420; US 2021285965 A1 20210916

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